



Genomic Unity[®] Testing

Tandem repeats analyzed

The following characterized repeats are detected and analyzed by the Genomic Unity[®] test.

Gene(s)	Repeat	Disorder
<i>AFF2</i>	CCG	Fragile XE syndrome
<i>AFF3</i>	CGG	FRA2A fragile site
<i>AR</i>	CAG	Spinal and bulbar muscular atrophy
<i>ATN1</i>	CAG	Dentatorubral-pallidoluysian atrophy (DRPLA)
<i>ATXN1</i>	CAG	Spinocerebellar ataxia
<i>ATXN10</i>	ATTCT	Spinocerebellar ataxia
<i>ATXN2</i>	CAG	Spinocerebellar ataxia
<i>ATXN3</i>	CAG	Spinocerebellar ataxia
<i>ATXN7</i>	CAG	Spinocerebellar ataxia
<i>ATXN80S</i>	CTG	Spinocerebellar ataxia
<i>C9ORF72</i>	GGGGCC	Frontotemporal dementia and/or amyotrophic lateral sclerosis (FTDALS1)
<i>CACNA1A</i>	CAG	Spinocerebellar ataxia
<i>CNBP</i>	CCTG	Myotonic dystrophy type II
<i>CSTB</i>	CCCCGCCCGCG	Myoclonus epilepsy
<i>DIP2B</i>	CGG	FRA12A fragile site
<i>DMPK</i>	CTG	Myotonic dystrophy type I
<i>FMR1</i>	CGG	Fragile X syndrome
<i>FXN</i>	GAA	Friedreich's ataxia
<i>HTT</i>	CAG	Huntington disease
<i>JPH3</i>	CTG	Huntington disease-like 2 syndrome
<i>NOP56</i>	GGCCTG	Spinocerebellar ataxia
<i>NOTCH2NLC</i>	GGC	Neuronal intranuclear inclusion disease
<i>PHOX2B</i>	Alanine	Congenital central hypoventilation syndrome
<i>PPP2R2B</i>	CAG	Spinocerebellar ataxia
<i>TBP</i>	Glutamine	Spinocerebellar ataxia
<i>TCF4</i>	CAG	Fuchs endothelial corneal dystrophy

Sensitivity and reporting policies differ by loci. The false negative rate for repeat expansions has not been determined for the following genes: *AFF2*, *AFF3*, *ATXN10*, *CNBP*, *CSTB*, *DIP2B*, *NOTCH2NLC*, *PHOX2B*, *TBP*.